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Hereditary Blindness.

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# HEREDITARY BLINDNESS

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ANY attempt to catalog the major causes of blindness cannot fail to include the classification "Hereditary." It is not the purpose of this paper to review the scientific researches which have been made from time to time to determine the number of those who are blind or who have suffered serious loss of sight due to this cause. Such estimates vary from ten to fifteen and even a greater percentage.

The late Dr. Lucien Howe of Buffalo devoted much of his time, especially during the later years of his life, to research in the field of hereditary blindness. He proposed and advocated legislative measures to prevent the marriage of any person blind from an inheritable cause; but as far as I can learn, no such legislation has ever been placed on the statute books.

The question of the reduction or elimination of the number of persons blind from heredity is perhaps the most perplexing problem of the movement for the prevention of blindness—much more complicated than the reduction of blindness from such other causes as trachoma, accidents, ophthalmia neonatorum, etc. As a matter of fact, such activities as society may undertake to bring about a reduction of blindness from this cause must of necessity be a part of a larger eugenics program. From the beginning of the present century there have been undertaken a vast number of genetic researches which grew out of the discovery of Mendel's laws of heredity. It has been definitely established by these researches that certain physical characteristics are inherited and are inevitable in any scheme of propagation either in the vegetable or animal kingdom.

In the case of all animal life, where mental processes are a part of existence, the same limitations are placed upon the embryo resulting from the union of the egg and the sperm, such embryo resulting from a selection in equal numbers of the chromosomes of the two uniting nuclei.

The laws of inheritance are fixed and immutable and any attempt to improve society such as the eugenists are striving to accomplish, must be based upon the conscious control of the future race through the elimination of the reproduction of the physically and mentally unfit. This is a big proposition and has many implications into which this paper cannot go. It is sufficient to say that it is perhaps the greatest problem of society at the present time.

Considered in many of its aspects the question of inherited blindness is relatively small compared with the handicaps upon society through inheritance of certain other physical and mental characteristics, such as insanity, feeblemindedness, and epilepsy. The aim of society is to produce individuals who can successfully adjust themselves to the environment in which they must live. In one respect at least the question of hereditary blindness does not have the sociological importance which the other inherited characteristics do have; that is, blindness does not of necessity, imply diseased or impaired mind, and, after all, the greatest handicap which an individual can have insofar as its resulting in a permanent inability to adjust to a given environment, is that of a high degree of feeblemindedness or insanity.

Again, considered with reference to the amount of inherited blindness in comparison with other inherited handicapping physical characteristics, blindness is relatively limited. The number of blind people from all causes in the United States is approximately one out of every 1,000, or one-tenth of one per

cent. Assuming that 10% of those who are blind are blind from an inheritable cause, we have one out of every 10,000 or one one-hundredth per cent of the total population so affected.

As a general proposition, modification of the human race through selective parenthood or through the exercise of will on the part of the potential parent is very hard to accomplish. The perpetuation of animal life is dominated by instinct rather than reason. Any such control through reason and deliberation presupposes a high degree of intelligence and understanding and control over instinctive forces and this is found in an almost negligible proportion of the human race. The attacks which eugenists have been making through the advocacy of legislative restrictions and through sterilization have so far been quite limited in their application and such as have taken place have been an attempt to prevent the propagation of those who will be beyond question drags on society. It is probable, as time goes on, that a greater and greater amount of attention must be given to this problem if there is to be conscious control of the quality of future generations. There is some evidence to show that public health and welfare activities and the accumulation of wealth have militated somewhat against the natural law of the elimination of the unfit, and that the proportionate number of unfit is greater now than ever before. However, it may be that with the growth of humanitarian ideals and practices the number of individuals who are receiving the attention of society because of their inability to adjust themselves is greater than ever before.

What has been said above is not intended to deprecate the importance of reducing the number of blind from inheritable causes, but rather to give it its proper setting in the whole problem of heredity insofar as that problem affects the future welfare of the human race. However, those who work for and with the blind, and the blind themselves, have a responsibility for efforts which will tend to reduce the amount of inherited blindness.

### Types of Hereditary Blindness

First of all, every blind person should know, insofar as he is able to ascertain, the exact cause of his blindness.

Dr. Clarence Loeb, of St. Louis, Missouri, prepared in 1909 the best statement that I have come across on hereditary blindness in his article on "The Marriage of the Blind from the Standpoint of a Physician." This paper gives the following principal forms of blindness which may be inherited:

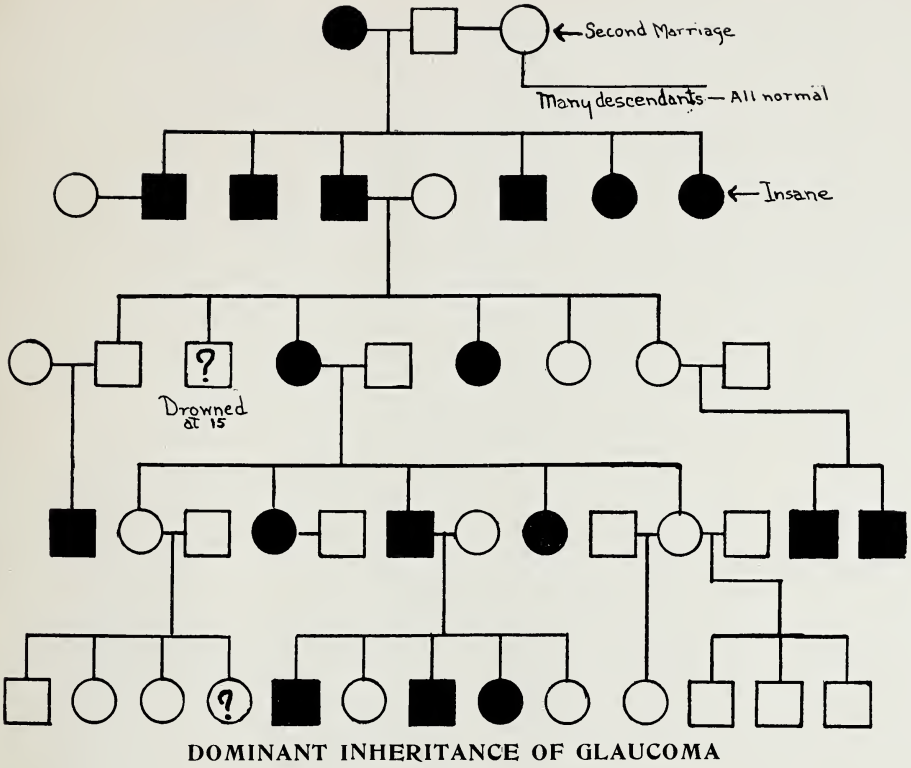
1. *Anophthalmus and Microphthalmus.*—That is to say, a total absence of the eye or an eye so small as not to provide useful vision. After a careful research, Dr. Loeb concluded that of such cases studied 74% of the offspring were affected through a direct, indirect, or collateral inheritance of this form of blindness.

2. *Buphthalmus.* This is the opposite of the foregoing and means an eye too large to focus. Here, again, the percentage of collateral inheritance exceeds 70%.

3. *Albinism,* or the absence or great shortage of pigment from the tissues of the eyes and especially in the iris which serves to shut out the excess of light. Dr. Loeb concludes that direct inheritance from this cause is only 12% but that collateral inheritance is very marked. In 8 families with 43 children he found 21 albinos, or 48%.

4. *Family Degeneration of the Cornea.* For this cause Dr. Loeb collected the histories of 18 families, 10 fathers and 8 mothers, with 62 children, of whom 26 were diseased, or 42%. Thirteen families with 50 children were reported, of whom 34, or 68%, showed collateral inheritance.

5. *Aniridia, and Coloboma Iridis.* This means the absence either wholly or in a large part of the iris. Dr. Loeb states that this condition shows marked direct heredity. The statistics which he gathered cover 59 families, with 29 affected fathers and 30 affected moth-




**DOMINANT INHERITANCE OF GLAUCOMA**

**Figure 7**

Chart showing inheritance of juvenile glaucoma simplex (Adapted from Courtney and Hill, *Journal of American Medical Association,* November 28, 1931). The family traces back to a Scotch immigrant, who arrived in Virginia about the middle of the Eighteenth century. He married a woman suffering from glaucoma, and the defect has appeared in every generation since that time. An accurate record has been kept of the members of this family who suffered from glaucoma. In spite of the fact that the hereditary nature of the disease was recognized, the families of glaucomatous individuals have been large. That this should be the case in spite of the painful and tragic consequences of the disease, is apparently due to the fatalistic religious beliefs of the members of this family, for they are obviously well above the average in intelligence and some of them have been leaders in the affairs of the nation in spite of the enormous handicap of this disease which has afflicted several members of each generation. They live now in an inaccessible foothill region of Virginia. What effect will be produced by a clear understanding of the mechanism of the inheritance of this tragic defect remains to be seen. The certainty that half the children of glaucomatous individuals will, on the average, be doomed to inherit this affliction, and that those without symptoms of glaucoma will not transmit it (unfortunately not always the case in this family if the record in the third generation is accurate) will serve both as a warning and an assurance.

ers; there were 156 children, of whom 116 were affected, making a perecntage of 74%.

6. *Ectopia Lentis.* In this eye affection the crystalline lens is displaced and as a result the rays of light entering the eye are unequally refracted and a disturbance of vision takes place. In this instance 3 cases of direct heredity were quoted, the taint coming from the father in 19 cases and the mother in 24 cases. There were 155 children, of whom 109 were affected, a percentage of 70%.

7. *Cataract.* Dr. Loeb says: "Even if the lens is in its proper position, it

may be opaque, so that the disturbance in vision ranges from very slight to only ability to distinguish between day and night, depending on the amount and location of the opaque spots. This is one of the most frequent forms of hereditary blindness. The cataract may appear very late in life, or may be congenital. In some cases, each succeeding generation is affected at an earlier age, a condition called anticipation." Inasmuch as cataract appears to be the most frequent cause of inherited blindness, it may be well to quote still further from Dr. Loeb, and such extended quotation will also give an example of the thoroughness of this whole investigation:

Berry reported a family where the history was known for seven generations. First generation was normal. There were 2 members to the second generation, both of whom were normal. In the third generation there were 10 children, of whom 5 had cataract. In the fourth generation there were 9 children, all of a cataractous mother, of whom 4 had cataract and 5 were normal. In the fifth generation there were 6 children, all of a cataractous mother, of whom 4 had cataract. In the sixth generation there were 10 children, descended as follows: A cataractous mother had 1 normal and 4 cataractous children; a cataractous father had 2 normal children; a cataractous mother had 1 normal daughter, and a normal mother had a normal son. The seventh generation had 21 children, as follows: A normal father had 10 normal children; a cataractous mother had 2 normal children and 1 cataractous daughter; a cataractous father had 1 cataractous daughter by his first wife and 3 normal children by his second; another cataractous father had 1 cataractous daughter and 2 normal children; a normal mother had 1 normal child. Cooper stated in 1852 that he had been informed by the Duke of Sussex that cataract had been brought into the royal family of England by the marriage of one of his ancestors with a princess of Saxo-Coburg-Gotha. She had cataract and many of her descendants had the same affection. Among them was the Duke of Cumberland, who commanded at Fontenoy; George III, George IV, the Duke of Gloucester, the Duke of Sussex, Princess Sophie, and the King of Hanover.

Including the cases reported in the letters received, I have succeeded in collecting the histories of 304 cases of direct heredity of cataract. The father was affected 145 times, the mother 152 times, not stated, 7 times. There were in all 1,012 children, of whom 589 had cataract, and 423 had normal eyes; percentage of affected children, 58. In other words, in every family of 5 children, where either the father or mother have cataract, 3 of the children will at some time suffer from cataract. The percentage would probably be higher were it not that so many people with cataract die young, and so leave no descendants.

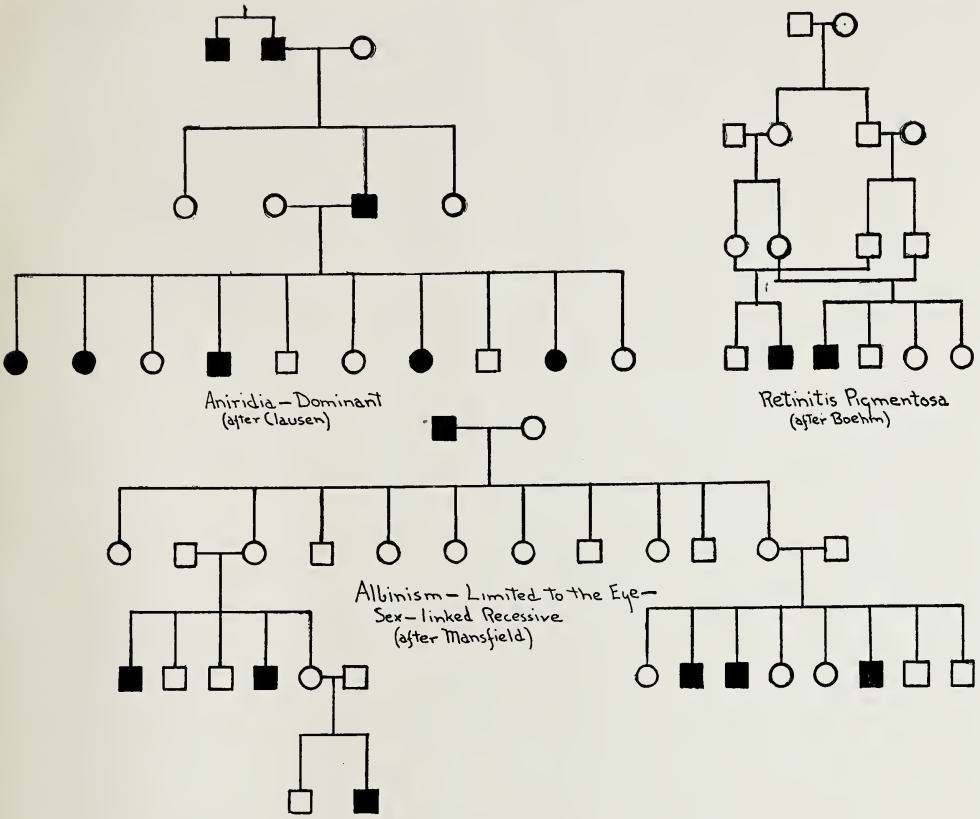
I have found only one case where both the father and the mother had cataract. There were 8 children, 4 of whom were normal, 2 had cataract, and 2 unknown. To these must be added the cases reported by Drs. ——— and ———. In one of these families there were 2 and in the other there were 5 children with cataract, in all 3 families with 15 children, of whom 9 had cataract, making 60%—slightly more than where only one parent is affected. The number of cases is too small, however, for a general rule.

I have found 13 cases of indirect inheritance from grandparent to grandchild, and 16 cases of indirect inheritance from uncle or aunt to nephew and niece. There were in these 29 families 107 children, of whom 62 were normal and 45 had cataract, equaling 42%.

In the literature, 66 cases have been reported of collateral inheritance; that is, cases of two or more children in the same family having cataract, while all other relatives have been free. There were 118 normal and 186 cataractous children. To these must be added the cases reported by Drs. ——— and ——— and ———, making 68 families, with 119 normal and 193 cataractous children, or 61.8% of the latter.

8. *Retinitis Pigmentosa.* This condition, which is practically the only disease of the retina showing hereditary tendencies, is characterized by a diminution of vision especially pronounced on a dark day and towards nightfall, although there may be almost total blindness on even bright days. This affection is subject to direct heredity, but its usual appearance is in the form of collateral inheritance, and it is often found in the offspring of consanguineous marriages. Of 133 cases of collateral inheritance reported, there were 541 children, of whom 349 were affected, or 64%. This is considerably larger than the incidence of direct inheritance, as shown by a record of 126 families, 61 fathers and 65 mothers, with 405 children, of whom 205, or 50%, had directly inherited the condition from the parent.

9. *Atrophy of the Optic Nerve.* This




**INHERITANCE OF OTHER FORMS OF BLINDNESS**

**Figure 8**

Three inheritance charts (modified from Crew) showing inheritance of Albinism limited to the eye, Aniridia (absence of iris), and retinitis pigmentosa. The Albinism is inherited as a sex-linked recessive, the retinitis pigmentosa as a recessive, and Aniridia as a dominant.

is a form of blindness due to the degeneration or death of the optic nerve even though the rest of the eye may be normal. It is usually due to syphilis, but may be an inheritable condition, when it usually occurs in the form of indirect heredity from uncle to nephew; females are said to be rarely affected, although their male children frequently are. In studies made of direct, indirect, and collateral inheritance of this affection, the incidence was respectively 46%, 60%, and 70%.

10. *Glaucoma.* This disease, which in acute cases is characterized by severe pain, is responsible for a more or less rapid loss of sight due to the effect of the disease upon the optic nerve. Not all forms are hereditary. In 44 families where there were 131 children, 72 of them, or 55%, showed direct hereditary glaucoma. In 13 cases of collateral inheritance, 33 out of 53 children were affected, or 62%.

11. *Nystagmus.* By this is meant a more or less continual movement of the eyes. The condition is hereditary. With 8 fathers and 10 mothers affected, 26 out of 56 children, or 46%, had the disease. In 9 cases of collateral inheritance, 29 out of 40 children were affected, or a percentage of 70%. Three

families had 11 children, or 100%, showing indirect heredity.

12. *Strabismus and Ophthmoplegia Externa and Ptosis.* Ophthmoplegia externa, which is a more or less complete paralysis of one or more of the muscles of the eye or eyelid, is usually directly hereditary. The records of 32 cases, with the father affected 24 times and the mother 8 times, showed 121 children, 77 of whom, or 63%, had inherited the condition. In five cases of collateral inheritance there were 17 children, of whom 13, or 76%, were affected.

The above extended references to Dr. Loeb's pamphlet indicate fairly well the type of research which has been conducted to arrive at the probability of inherited eye defects. Some of the causes which are given are the result of syphilis, which is not mentioned by Dr. Loeb as a cause of hereditary blindness. It appears to me that considerable research still needs to be undertaken to determine eye defects which are the result of inherited syphilis. It is not possible within the limits of this paper to enter into any discussion of the question of syphilis as an inheritable disease, or to point out the distinction which can be made between certain forms of eye trouble which persist through many generations which are distinct from those caused by this dread disease.

But when it comes to the question of the responsibility for future generations, every person who is affected with this disease, whether he acquired it through heredity or any other means, must take the same and additional precautions as have been suggested above as the responsibility of anyone who has lost his sight as the result of an inherited physical characteristic.

It has not generally been an activity of organizations interested in the prevention of blindness to engage in a first-hand attack on the eradication of syphilis, although the extent of eye havoc which results from that disease is sufficiently extensive to warrant all possible support in the war for its extermination.

### Non-Inheritance of Acquired Blindness

There are still many beliefs in some quarters which are fallacious and are based upon the wrong assumption that an acquired physical characteristic can be inherited. Therefore, any one who is blind from such a cause as trachoma, gonorrheal ophthalmia, or accident need have no fear of marriage from the standpoint of transmitting his lack of sight.

There is also one other confusion of terms sometimes made, that is the difference between congenital blindness and hereditary blindness. Not all here-

TABLE I.—Incidence of Blindness in the Children of the Blind*

| | *Disease* | *Families* | *Total Children* | *Affected Children* | *Normal Children* | *% Affected Children* |
|---|---|---|---|---|---|---|
| I. | Albinism | 13 | 60 | 23 | 37 | |
| II. | Aniridia and Colomba Iridis | 72 | 199 | 147 | 52 | 73.8 |
| III. | Anophthal and Microphthal | 48 | 117 | 87 | 30 | 74.4 |
| IV. | Atrophy of Optic Nerve | 150 | 506 | 312 | 194 | 61.6 |
| V. | Cataract | 404 | 1446 | 836 | 610 | 57.8 |
| VI. | Ectopia Lentis | 64 | 212 | 155 | 57 | 73.1 |
| VII. | Family Degen. of the Cornea | 32 | 114 | 62 | 52 | 54.4 |
| VIII. | Glaucoma | 58 | 194 | 109 | 85 | 56.2 |
| IX. | Megalophthalmus | 8 | 28 | 21 | 7 | |
| X. | Nystagmus | 30 | 107 | 66 | 41 | 61.7 |
| XI. | Ophthalmoplegia and Ptosis | 39 | 145 | 94 | 51 | 64.8 |
| XII. | Retinitis Pigmentosa | 286 | 1027 | 611 | 416 | 59.5 |
| | Total—All Diseases | 1204 | 4155 | 2523 | 1632 | 60.8 |

*Based on table appearing on page 38 of "The Marriage of the Blind from the Standpoint of a Physician," by Dr. Clarence Loeb.

ditary blindness or eye defects are congenital; for example, the most common kind of cataract is in many instances an inherited defect which does not appear until middle or late life. In some instances blindness may be both congenital and hereditary, as for example anaphthalmos, or the total absence of the eye. On the other hand, there is a considerable amount of congenital blindness which is not hereditary. This is due to accidents of birth. Fortunately, this is being appreciably reduced due to the great advancement which has been made in obstetrical practice.

Nature provides immunity to the embryo from practically all communicable diseases with the exception of syphilis, and any disease acquired by the child in the process of birth cannot be classified as inherited. Ophthalmia neonatorum, so often erroneously given as blindness from congenital causes, is communicated to the eyes of the child at or after birth, and hence cannot be classified as congenital.

This paper has attempted to deal exclusively with the subject of the sociological aspects and implications of hereditary blindness and has not entered into that larger question of the economic status of the afflicted. This is a general consideration which must be solved by everyone, including those who have lost their sight.

## The Genetics Congress

**Ithaca, New York, August 24-31**

THE preliminary program of the Sixth International Genetics Congress has just been issued (August 1). This adds considerably to the details of the statement regarding the Congress published in the July JOURNAL, but time is so short that it is hardly worth while to publish the program in full. Readers of the JOURNAL who are planning to attend the Congress will have access to the final program by the time that this appears, including the abstracts of all papers and demonstrations to be presented at the Congress. Those who are not going to Ithaca will be more interested to learn of the high points of the Congress in our September number.

In spite of adverse economic conditions, it is certain that the Congress will be truly international in character. Between fifty and seventy-five delegates from Europe have signified their intention of attending, with representation from Japan and other countries also assured. The total enrollment is at present well over seven hundred.

At the five general morning sessions of the Congress, the following topics will be discussed by world authorities: *Thursday, August 25,* General Genetics; *Friday,* Mutations; *Saturday,* the interrelations of Cytology and Genetics; *Monday,* Genetics of species Hybrids; *Tuesday,* Contributions of Genetics to the Theory of Organic Evolution. The speakers who have accepted places on these programs were given in the announcement in the July number.

Our three hundred geneticists from thirty countries are taking part in the program, either with papers or with exhibits or demonstrations. Section meetings will be devoted to the following subjects: General Genetics; Cytology; Animal Genetics; Human Genetics; Methods and Technique; Genetics and Phytopathology; Chromosome structure and Crossing Over; Genetics of Species Hybrids; Drosophila; Genetics and Pathology; Problems Relating to Sex and Fertility; Fruit and Vegetable Breeding. The entire range of genetic research will be covered by the exhibits and demonstrations. These, and the social events, for which time has ingeniously been found by the committees including arrangements, should make the week one long to be remembered.

# MUTTON, AND HOW IT GETS THAT WAY

## A Review

GROWTH AND THE DEVELOPMENT OF MUTTON QUALITIES IN THE SHEEP, *by* JOHN HAMMOND. 597 pp. + XXVI, 139 tables (exclusive of the appendix), 137 figures, 71 diagrams and 2 text figures. Edinburgh and London, Oliver and Boyd. 1932.

"The object of the present investigation was to make a general survey of the scientific principles involved in the production of meat from the physiological, anatomical and practical points of view."—Introduction.

"As has been pointed out before, the object of the work was a general survey of the principles involved rather than the conclusive proof of the correctness of one or two of them, its aim being the suggestion of profitable lines of experimental work bearing on the development of the animal from the meat point of view."—page 198.

MR. HAMMOND has produced a compendium of information on how the sheep becomes what it is. The data presented are the author's own and the point of view is original and stimulating. Many of the topics discussed are not new but because of the author's original method of attack and original data, he comes to new conclusions with which the reader may not agree but which he can not ignore.

An extensive review of other works is evident in the number of titles cited. Part I, which deals with changes in live weight, ends with a list of 118 references. Part II, about the weights of the carcass and the organs, mentions only 77 titles. Part III, the various proportions of the skeleton and the effects of these on conformation, ends with the listing of 175 other works. Part IV, variations in the proportions of muscle, fat and bone in the carcass, is attended by 117 references. Part V (written in conjunction with A. B. Appleton) is a detailed study of the leg of mutton and includes an especially noteworthy section on the histoloy of the muscles. There are 173 references.

The comments on other works are particularly well arranged, being scattered under the appropriate subjects and along with discussion of the author's own findings, instead of being placed all together in a formal "review of literature," or "previous works" as is sometimes done. The reviewer is inclined to regret that the author has not been more critical in his references to these other works. In some places he seems to have cited with almost equal faith and enthusiasm the statements of those who were reporting their own actual data and those who were reporting hearsay; of those who wrote 50 years ago in the light of the science of their time, and those who wrote in the last ten years in the supposedly brighter light of twentieth century science; of those who worked with one or two animals and those who worked with dozens.

The book is monumental in scope. It is almost unique in the enormous number of questions which it attempts to answer. As an authoritative source-book its most serious weakness lies in the small amount of data bearing on any question except in Part I. Many tables in Parts II, III, IV, and V contain no averages based on more than three individuals. For example Table 36, on the proportions of the carcass and organs in different breeds, contains data on *only 10 individuals* and these are scattered among *six different breeds.* Within breeds this scant material was hardly even comparable: two of the four Suffolks are wethers while two are rams. Throughout the entire book the conclusions about the effects of domestication are mostly based on the data from one Soay and one Shetland ram. What a terrific responsibility to place upon two rams differing in breed and age, that of being truly representative of wild sheep in general!

Where the numbers in each group are as small as they are in most tables in this book, and especially where each group is used as evidence on two